Down Syndrome

BIRTH DEFECT RESEARCH FOR CHILDREN

What is Down Syndrome?

Down syndrome (DS) is a chromosomal abnormality due to excess genetic material found in the genes along the 21st chromosome. Since 95% of all DS cases occur because there are three copies of the 21st chromosome rather than the usual two, it is often referred to as trisomy 21. The additional genetic material alters the course of development and causes the characteristics associated with DS.

Down Syndrome

Symptoms include:

varying degrees of mental retardation
flattened facial features
an upward slant to the eyes
skin folds at the inner corners of eye (epicanthal)
short limbs, hands, and fingers
microcephaly (small head)
low muscle tone (muscle hypotonia)
Simian crease (single deep crease on palm)
hyperflexibility

How many children are born with Down Syndrome?

Down syndrome is one of the most frequent chromosomal abnormalities found in humans, occurring approximately once in every 800 live births. That is 5,000 babies each year. Over 350,000 people, in the United States are affected by DS. Women 35 and older are at an increased risk (1 in 400) of having a child with DS. The risk increases to 1 in 35 births for mothers over 45. However, due to the higher fertility rates, around 80% of children with Down syndrome are born to mothers under the age of 35.

What causes Down Syndrome?

There are three different types of chromosomal abnormalities that may lead to DS:

Non-disjunction is the most common error that takes place during cell division occurring in 95% of all DS cases. Non-disjunction is the failure of two corresponding chromosomes to separate during the first division of meiosis. As a result, an embryo will contain three number 21 chromosomes instead of two. The embryo will continue to develop, replicating an extra chromosome in every cell of the body.

Translocation occurs in 3 to 4 % of people with DS. Part of the number 21 chromosome breaks off during cell division and attaches to another chromosome. The total number of chromosomes in this case remains 46, but the extra part of the 21st chromosome causes the features of DS.

Mosaicism occurs when nondisjunction takes place after fertilization in one of the initial cells. In this case some cells have 46 chromosomes and some have 47. The cells with 47 chromosomes have an extra 21st chromosome. Mosacism is very rare, accounting for 1 to 2 percent of all cases of DS.

Prenatal tests:
There are screening tests and diagnostic tests to determine whether an unborn baby may have Down Syndrome.

Screening tests:
Triple Screen & Alpha-fetoprotein Plus are the most common screening tests used. They combine information about the mother (age, etc.) and substances found in the blood to estimate the risk of having a fetus with DS. These screening tests can accurately detect 60 % of DS cases.

Diagnostic tests:
Chorionic Villus Sampling (CVS), amniocentesis and percutaneous umbilical blood sampling (PUBS) are diagnostic tests that are about 98-99 % accurate in the detection of DS. Each of these procedures carries a small risk of miscarriage. CVS has also been associated with an increase in certain birth defects in some studies.

Caring for a child with Down Syndrome

Medical:
Children with DS are at an increased risk for certain health problems. The most commonly associated problems are congenital heart defects, greater susceptibility to infection, respiratory problems, obstructed digestive tracts and childhood leukemia. Life expectancy for people born with DS has increased dramatically due to the advance-ment in medicine and is now approximately 55 years.

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Education: Parents of a child with DS should look into early intervention/infant development programs as soon as possible.

These programs can educate parents about the proper strategies to help their child develop to his/her fullest potential. Depending on the severity of mental retardation, many people with DS can be educated and live independent lives.

Future: The opportunities available to people with DS today have never been greater. Independent Living Centers, group-shared and supervised apartments and support services in the community have proven to be important resources for the independence of people with disabilities.

Fact Sheet by:

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